ABSTRACT

The present invention relates generally to the field of human genetics. Specifically, the present invention relates to methods and materials used to isolate and detect a human prostate cancer predisposing gene (*HPC2*), some alleles of which cause susceptibility to cancer, in particular prostate cancer. More specifically, the present invention relates to germline mutations in the HPC2 gene and their use in the diagnosis of predisposition to prostate cancer. The invention also relates to presymptomatic therapy of individuals who carry deleterious alleles of the *HPC2* gene. The invention further relates to somatic mutations in the *HPC2* gene in human prostate cancer and their use in the diagnosis and prognosis of human prostate cancer. Additionally, the invention relates to somatic mutations in the *HPC2* gene in other human cancers and their use in the diagnosis and prognosis of human cancers. The invention also relates to the therapy of human cancers which have a mutation in the *HPC2* gene, (including gene therapy, protein replacement therapy, protein mimetics, and inhibitors). The invention further relates to the screening of drugs for cancer therapy. The invention also relates to the screening of the HPC2 gene for mutations, which are useful for diagnosing the predisposition to prostate cancer. In addition, the invention relates to a paralog of human *HPC2*, the paralog being named *ELAC1*, and to orthologs of human *HPC2*, these being mouse *Elac2*, chimpanzee *Elac2* and gorilla *Elac2*.